Docket No.: HO-P02086US1
(PATENT)

IN THE UNITED STATES PATENT AND TRADEMARK OFFICE

In re Patent Application of:
James R. Lupski, et al.

Application No.: 10/021,955 Group Art Unit: 1637

Filed: December 13, 2001 Examiner: S. Chunduru

For: DEFECTS IN PERIAXIN ASSOCIATED WITH MYELINOPATHIES

DECLARATION UNDER 37 CFR §1.132

Dear Sir:

I, James R. Lupski, M.D. Ph.D., do hereby depose and say as follows:

1. I am a United States citizen residing at 11102 Ashcroft Dr, Houston, TX, 77096, USA.

2. I am an employee of the assignee of the above-referenced patent application, I am an inventor of said application, and I have read the contents of said application.

3. I am a Professor of Molecular and Human Genetics at Baylor College of Medicine in Houston, Texas. I am skilled in the area of molecular genetics and disease. A resume describing my experience is attached to this declaration.

The Examiner is alleging in the outstanding Office Action that the presently pending claims are rejected under 35 U.S.C. §112, first paragraph, as not being enabled by not teaching how to make and use the invention commensurate with the scope of the claims. This is an inaccurate assessment of the nature of the present invention and the technology involved for this particular application.

The Examiner alleges that the specification does not teach that a statistically significant correlation exists between all *PRX* mutations and any myelinopathy or that a predictable correlation can be made as to an association between any *PRX* mutation and any myelinopathy. However, we have demonstrated a sufficient correlation between several *PRX* mutations and a spectrum of highly related demyelinating neuropathies, and therefore those of skill in the art would certainly consider there to be a correlation. For example, we have shown in at least paragraphs **[0244]** and **[0260]** of the specification that several mutations,

including 2787ΔC, 2857C>T, and so forth, are the cause of autosomal recessive DSN. Furthermore, we show in at least paragraphs **[0264]**, **[0268]** and **[0273]** that mutations 2145T>A and 247ΔC cause a broad spectrum of demyelinating neuropathies, including CMT myelinopathies and DSN.

The invention does not concern mutations in *PRX* for a wide range of non-related diseases but rather concerns those *PRX* mutations as part of a phenotypically narrow range of myelinopathies. That is, there is considerable phenotypic overlap among the myelinopathies, as discussed in the specification (see paragraphs **[0074]** through **[0083]**), including at least defects in myelin, but also onion bulb defects (found in CMT1, DSS, and CHN); slowed motor nerve condution velocities (NCV) (found in CMT1, HNPP, DSS, and CHN); muscle weakness (CMT1 and CHN); gait disturbance or ataxia (CMT1 and RLS); and areflexia (CHN and RLS), for example.

Thus, a skilled artisan recognizes that myelinopathies such as Dejerine-Sottas neuropathy (DSN) and Charcot-Marie-Tooth disease type 1 (CMT1) are only part of a spectrum of neuropathy phenotypes having different degrees of severity. Although at least fifteen genetic loci and six genes have been associated with this spectrum of disorders, this does not preclude a single gene such as *PRX* from having a mutation in each of these related disorders for a subset of patients and families. In fact, there is a precedent for there being more than one defective gene even within the family itself, given that for CMT1, related mutations in addition to *PMP22* include those in *MPZ*, *Cx32*, *EGRα*, *MTMR2*, *NDRG1* (see paragraphs **[0079]** and **[0080]**). Furthermore, DSS and CHN can be caused by mutations in multiple genes (*MPZ* and *EGR2* for both), and the specification even states (paragraph **[0080]**): "...these myelinopathies appear to represent a *spectrum of related disorders resulting from myelin dysfunction*". Therefore, it is clearly within the scope of the present invention to provide for mutations in a single locus, *PRX*, as diagnostic of a spectrum of myelinopathies. Furthermore, since the time of our original publication, other groups and we have confirmed the role of *PRX* in myelinopathies.

The Examiner also expresses concern about the unpredictability of identifying whether a sequence variation is a polymorphism or a disease-causing mutation, but in diseases such as those myelinopathies that comprise an autosomal recessive nature (see, at least, paragraphs **[0062]**, **[0242]**, **[0244]**, **[0246]**, **[0247]**, **[0260]**, **[0261]**, **[0268]**, and **[0273]**),

it is significant that mutations on both alleles must be present before the disease occurs, whether as a homozygote or compound heterozygote. That is, it is highly unlikely with an affected family that two non-diseased parents of a diseased individual would be carriers of the same polymorphism. In fact, if the myelinopathy had an inheritance pattern other than autosomal recessive, we would be able to easily identify this, as well.

Moreover, a skilled artisan recognizes how to discern between a polymorphism and a disease-causing mutation. If the sequence alteration is a polymorphism, it is not identified in controls and/or does not segregate with the disease phenotype. By definition in the field of human genetics a polymorphism has to be observed in 1% of chromosomes. Thus, the absence of such a variant in 50 control normal individuals (*i.e.* 100 control chromosomes) is inconsistent with the variant representing a polymorphism.

Furthermore, new claims 43-49 have been added and are directed to myelinopathies that comprise prominent *sensory* neuropathies. One of skill in the art recognizes based on the knowledge in the art (Takashima et al., 2002) and the teachings of the specification (paragraphs **[0268]** and **[0273]**) that *PRX* mutations associate with prominent *sensory* neuropathies. That is, whereas defects with many Hereditary Motor and Sensory Neuropathies (HMSN) have significantly stronger motor defects, those that are the result of *PRX* mutations have significantly stronger sensory defects. Thus, in new claims 43-39 Applicants are highlighting those myelinopathies that have an oppositely manifesting phenotype from myelinopathies arising from mutations in genes such as *PMP22*, *MPZ*, and so forth.

4. I hereby declare that all statements made herein on my own knowledge are true and that all statements made on information and belief are believed to be true; and further that these statements were made with the knowledge that willful false statements and the like so made are punishable by fine or imprisonment, or both under Section 1001 of Title 18 of the United States Code and that such willful false statements may jeopardize the validity of the application or any patent issued thereon.

Date: Dec. 12 2003 James R. Lupski

James R. Lupski, M.D. Ph.D., FAAP, FACMG, FAAAS

December 10, 2003

CURRICULUM VITAE

NAME: James R. Lupski

BIRTHDATE: February 22, 1957; Hicksville, New York

CITIZENSHIP: U.S.A.

MARITAL STATUS: Married to Gabriella R. Gerardi, June 14, 1986

CHILDREN: Alessandra Marie Lupski, Born: July 27, 1989

Marcella Dianne Lupski, Born: January 20, 1993

Joseph John Lupski, Born: August 19, 1998

SOCIAL SECURITY NO.: 115-52-5451

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Baylor College of Medicine
One Baylor Plaza, Room 609E
Houston, Texas 77030

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(713) 798-3723 Admin Coord (Betty Fernandini)
(713) 605-8989 15773 (beeper)
(713) 728-0344 home
Email: jlupski@bcm.tmc.edu

WEB SITE: **http://www.imgen.bcm.tmc.edu/molgen/lupski/**
http://www.imgen.bcm.tmc.edu/molgen/lupski/sms/Index-SMS.htm

EDUCATION:

DEGREE	INSTITUTION	DATE AWARDED	FIELD
B.A.	New York University	1979	Chemistry and Biology
M.S.	New York University	1983	Biochemistry
Ph.D.	New York University	1984	Biochemistry/Molecular Biology
M.D.	New York University	1985	Medicine

CLINICAL TRAINING:

Residency	Baylor College of Medicine	1986-1989	Pediatrics
Fellowship	Baylor College of Medicine	1989-1991	Medical Genetics

PROFESSIONAL MEDICAL LICENSURES:

New York State Medical License (license number: 177352; February 7, 1989-present; inactive)

Texas State Medical License (license number: H5185; February 24, 1989-present; active)

BOARD CERTIFICATIONS:

National Medical Board Delegate, 1986

Board Certified: Diplomate of the American Board of Pediatrics, 1989-1996
-Recertification, American Board of Pediatrics, 1997-2003

Fellow of the American Academy of Pediatrics, 1991-present
(Member of Genetics Section, AAP)

Board Certified: Diplomate of the American Board of Medical Genetics, 1993-2003
- Clinical Geneticist
- Clinical Molecular Geneticist
- Recertification, American Board of Medical Genetics, 2002-2012

Fellow of the American College of Medical Genetics, 1994-present

RESEARCH INTEREST:

Determine the molecular mechanisms for disease using human genetic approaches to investigate clinical phenotypes.

PROFESSIONAL EXPERIENCE:

Cullen Professor of Molecular and Human Genetics, and Professor of Pediatrics, Baylor College of Medicine, 1995-present.

Adjunct Appointment as Associate Professor (1993-1995), and as Professor (1995-present), Department of Microbiology and Molecular Genetics, The University of Texas Health Science Center at Houston.

Associate Professor with tenure (1992-1995), Assistant Professor (1989-1992), Research Assistant Professor (1986-1989), Institute for Molecular Genetics (Dr. C. Thomas Caskey, Chairman) (Department of Molecular and Human Genetics as of January, 1994; Dr. Arthur L. Beaudet, Chairman), and Department of Pediatrics (Dr. Ralph D. Feigin, Chairman), Baylor College of Medicine.

Attending Pediatrician and Medical Geneticist: Texas Children's Hospital, and Ben Taub General Hospital, Houston, TX 1991-present.

Consulting Geneticist: St. Luke's Episcopal Hospital, Texas Women's Hospital, The Methodist Hospital, Veteran's Administration Hospital and St. Joseph's Hospital, Houston, TX 1991-present.

Graduate Faculty Member in Neurobiology of Disease Graduate Program, Baylor College of Medicine, 1995-present.

Graduate Faculty Member in Graduate Program in Cell and Molecular Biology, Baylor College of Medicine, 1989-present.

Visiting Physician, Indian Health Service, Fort Defiance Indian Hospital, Fort Defiance Arizona, October, 1988.

Guest Professor, Center for Advanced Molecular Biology, University of the Punjab, New Campus, Lahore, Pakistan, February 1986

Research Assistant Professor, Faculty position in the Biochemistry Department (Dr. G. Nigel Godson, Chairman), at New York University Medical Center, 1985-1986.

Medical Scientist in Training with Professor G. Nigel Godson, New York University Medical Center, Biochemistry Department, 1979-1985.

Graduate Student with Dr. Ahmad I. Bukhari, Cold Spring Harbor Laboratories, Summer 1979.

Teaching Fellow, Chemistry Department, New York University, 1978-1979.

Undergraduate Research Participant (URP) with Dr. Ahmad I. Bukhari, Cold Spring Harbor Laboratories, Summer 1978.

SPECIAL COURSES:

Participant, Cold Spring Harbor Laboratory Course "Introduction of Macromolecules into Mammalian Cells." Instructors: Mario Capechi and Richard Mulligan, Summer 1982.

Participant, Cold Spring Harbor Laboratory Course "Genetic Approaches to Human Disease Using DNA Markers." Instructors: Eric Lander and David Page, Summer 1990.

Participant, The Johns Hopkins Medical School and The Jackson Laboratory Course "The 38th Annual Short Course in Medical and Experimental Mammalian Genetics." Organizers:
V. McKusick, D. Valle, J. Naggert and P. Nishina. July 1997.

Participant, Rice University "Advances in Tissue Engineering." Organizer: Antonios G. Mikos, August 2001.

POSITIONS HELD:

Medical Director, Bacterial BarCodes, Inc., 2000-present. (http://www.bacterialbarcodes.com)

European Molecular Genetics Quality Network (EMQN) External Quality Assessment (EQA) Scheme organisers, expert assessors and best practice organisers. 2000-2002.

International Advisory Committee, International Symposium on Signal Transduction in Health and Disease (STADY). Sponsored by N.I.H., U.S.A. and Tel Aviv University, Israel. 2000-2002.

E. Mead Johnson Award Selection Committee, Society for Pediatric Research, 2000-2002.
Editorial Committee *The Health Channel*™ (Genetics Section), Baylor College of Medicine. 1997-1998.

Moderator for Medical Genetics modules of Cyberounds™ [interMDnet Corporation], internet address (http://www.cyberounds.com), 1996-present.

March of Dimes Birth Defects Foundation, Scientific Review Committee. 1997-2002.

Director, National Eye Institute predoctoral training grant, 1994-2000.

Director, 1993-present; Co-Director, 1990-1993.
National Institutes of Health Medical Scientist Training (M.D.-PhD.) Program at Baylor College of Medicine.

Participant, SCIENCE-BY-MAIL Program of Houston Children's Museum.
This program pairs a scientist with a grammar school class to guide students through experiments and their interpretation, 1993-1995, 1999.

Board of Directors

Bacterial BarCodes, Inc. 2000-present.

American Society of Human Genetics, 2000-2002

Scientific Advisory Boards

Bacterial BarCodes, Inc. 2000-present.

Spectral Genomics, Inc., Chairman, 2003-present.

BCM (Baylor College of Medicine) Technologies, Inc. 1999-present.

The Wills Foundation, 1997-2001.

Editorial Boards

The American Journal of Human Genetics, 1995-1997
Neuron, 1997-2000
Neurogenetics, 1997-1999; American Editor 1999-present
Genomics, Associate Editor, 2000-present

Professional Advisory Boards

The Monarch School for gifted and challenged children, Houston, Texas. 1997-present.

Arbor School for children with special needs, Houston, Texas. 2002-present.

Medical Advisory Boards

Familial Dysautonomia Foundation, 2001-present.

Medical and Research Advisory Board PRISMS
(Parents and Researchers Interested in Smith-Magenis Syndrome), 1993-present.

Medical and Research Advisory Board, CMT International, 1990-present.
Medical Advisory Board, CMT Association
(formerly the National Foundation for Peroneal Muscular Atrophy [NFPMA]), 1990-present.

Advisory Board for Inborn Errors of Metabolism, King Faisal Specialist Hospital and Research Centre, Riyadh, Saudi Arabia, 1995.

CONSULTANT:

Athena Diagnostics (formerly, Genica Pharmaceuticals Corporation); Worcester, MA., 1993-present.

Council of Healthcare Advisors

The Frischer-Dambra Corporation.

SCIENTIFIC COFOUNDER:

Bacterial BarCodes, Inc. (Incorporated May 1999)
The company provides DNA fingerprinting of microorganisms for strain identification and molecular epidemiological studies. BBCI applies the patented rep-PCR technology and also provides access to databases of DNA fingerprints from important bacterial species and strains.

CURRENT GRANTS:

National Institute of Child Health and Human Development. P01 HD39420
Principal Investigator for project entitled "Chromosome Rearrangements and Mental Retardation."
Project I "Molecular Mechanism of Recurrent Rearrangements" – P.I.: Bernice Morrow
Project II "Molecular Mechanism for Smith-Magenis Syndrome and its Recombination Reciprocal" – P.I.: James R. Lupski
Project III "Telomere Integrity of *de novo* Terminal Deletions" – P.I.: Lisa G. Shaffer
Project IV "Evolution of Primate Genome Sequence Resulting in Susceptibility to Chromosome Rearrangements" – P.I.: David Nelson
Administrative Core – P.I.: James R. Lupski
FISH Core – P.I.: James R. Lupski; Co-P.I.: Pawel Stankiewicz
DNA Sequencing Core – P.I.: David Nelson
Funded 04/01/01 – 03/31/06
Direct Costs: $4,396,304

National Institute of Neurological Disorders and Stroke R01 NS27042
Principal Investigator for project entitled, "CMT peripheral neuropathy: IV. Genes and Pathogenesis."
Funded 01/01/02 - 12/31/05
Direct Costs: $1,344,349

National Eye Institute R01 EY13255
Principal Investigator for project entitled "*ABCR*, macular dystrophies and degeneration"
Funded 11/1/01 – 10/31/06
Direct Costs: $1,717,762

National Institute of Dental and Craniofacial Research
Principal Investigator for project entitled "Molecular basis of the craniofacial anomalies in SMS"
Funded 02/01/03 – 01/31/07
Direct Costs: $1,000,000

National Eye Institute R01 EY11780
Principal Investigator: Bassem A. Bejjani; Co-Investigator: James R. Lupski for project entitled, "A genetic approach to the role of *CYP1B1* in PCG"
Funded 12/01/00 - 11/30/05
Direct Costs: $1,250,000

March of Dimes Birth Defects Foundation
Principal Investigator for Project entitled, "Identification of Genes Responsible for Bardet-Biedl Syndrome"
Funded 06/01/01 – 05/31/04
Direct Costs: $257,292

National Institutes of General Medical Sciences, 2T32GM07330-24
Program Director for Medical Scientist Training Program
Funded 07/01/00 - 06/30/05
Direct Costs: $3,741,510

John B. Carter Foundation
Principal Investigator for project entitled, "Fingerprinting bacteria using repetitive sequence based PCR"
Funded 07/01/02 – 06/30/04
Direct Costs: $40,000

Current direct cost funding to the laboratory (exclusive of training grants) = $10,005,707

Previous awards total 30 grants including 9 from the National Institutes of Health (3 NINDS, 2 NEI, 1 NCI, 1 NICHD, 1 NIGMS, and 1 BRSG), 6 from the Muscular Dystrophy Association, 3 from the Foundation Fighting Blindness, 1 from the Steinbach Foundation, 2 from Abbott Laboratories, 1 from Ross Laboratories, 1 from the Pew Scholars Program, and 1 from the National Science Foundation with a total direct cost = $9,907,931.

PATENTS, INVENTIONS, AND COPYRIGHTS:

1. "Antisense oligonucleotide antibiotics complementary to the macromolecular synthesis operon, methods of treating bacterial infections and methods for identification of bacteria"
   Filed with United States Patent and Trademark Office: July 5, 1988
   Serial No.: 215,135
   Inventor: James R. Lupski
   Australia Patent N . 633495; term is sixteen years from July 5, 1989
   Continuation-in-Part (C.I.P.) filed: August 23, 1990
   Serial No.: 07/572,191
   Inventors: James R. Lupski and Leonard Katz (Abbott Laboratories)

United States Patent N . 5,294,533; term is seventeen years from March 15, 1994.
Filed with European patent Office: June 30, 1989; priority claimed July 5, 1988
Application No. 89909671.3
European Patent No. 0424473; issued May 8, 1996, term is twenty years from filing date of June 3, 1989
Canadian Patent No. 1,340,796; issued October 19, 1999
Canadian Patent No. 2,048,450; issued June 13, 2000
Licensee: Abbott Laboratories, Abbott Park, IL.

2. "Molecular diagnosis of autosomal dominant Charcot-Marie-Tooth disease"
Filed with United States Patent and Trademark Office: June 6, 1991
Serial No.: 07/711,615
Inventors: James R. Lupski, Pragna I. Patel, Roberto Montes de Oca-Luna, Odila Saucedo-Cardenas
United States Patent No. 5,306,616; term is seventeen years from April 26, 1994.
Licensee: Athena Diagnostics (formerly Genica Pharmaceuticals), Worcester, MA.

3. "Fingerprinting bacterial strains using repetitive DNA sequence amplification"
Filed with United States Patent and Trademark Office: October 23, 1991
Serial No.: 08/248,848
Inventors: James R. Lupski, James Versalovic, Thearith Koeuth
United States Patent No. 5,523,217; term is seventeen years from June 4, 1996
Continuation-in-Part (C.I.P.) filed August 24, 1993
United States Patent No. 5,691,136; term is seventeen years from November 25, 1997
European Patent No. 0610396; issued January 17, 2001, term is twenty years from filing date October 21, 1992
German Patent No. 69231646.9-08; issued January 17, 2001
Licensee: Bacterial BarCodes, Inc.

4. "Peripheral myelin protein coding sequence and method"
Inventors: Pragna I. Patel and James R. Lupski (Baylor College of Medicine), Ueli Suter, G. Jackson Snipes, Marino DeLeon, and Eric Shooter (Stanford University), Andrew Welcher (Amgen)
United States Patent No. 5,599,920; term is seventeen years from February 4, 1997
Licensee: Amgen, Inc., Thousand Oaks, CA. (Therapeutics)
Athena Diagnostics, Worcester, MA. (Diagnostics)

5. "DNA diagnosis of Hereditary Neuropathy with Liability to Pressure Palsies (HNPP) using CMT1A-REP probes"
Continuation-in-Part (C.I.P.) filed: September 30, 1993
Serial No: 08/129,902
Inventors: James R. Lupski, Liu Pentao, Benjamin B. Roa, Nacer E. Abbas, Pragna I. Patel
United States Patent No. 5,780,223; term is seventeen years from July 14, 1998.
Licensee: Athena Diagnostics, Worcester, MA.

6. "Methods for screening for agents that delay a cell cycle and a composition comprising an analogue of wild-type Era"
Filed with United States Patent and Trademark Office: August 20, 1996
Inventors: James R. Lupski, Robert A. Britton (Baylor College of Medicine), Donald L. Court, Bradford S. Powell (ABL-Basic Research Program, National Cancer Institute)
United States Patent No. 6,132,954; term is seventeen years from October 19, 2000.
Licensee: Pending

7. "Nucleic acid and amino acid sequences for ATP-binding cassette transporter and methods of screening for agents that modify ATP-binding cassette transporter"
   Filed with United States Patent and Trademark Office: February 27, 1997
   Inventors: Mike Dean, Rando Allikmets (NCI-FCRDC);
   Mark Leppert, Nanda Singh (University of Utah);
   Richard A. Lewis, James R. Lupski, Noah F. Shroyer,
   Yixin Li, Kent L. Anderson (Baylor College of Medicine);
   Jeremy Nathans, Hui Sun, Amir Rattner,
   Philip Smallwood (Johns Hopkins University School of Medicine)
   Licensee: Merck and Company, Inc., Rahway, N.J.
   Active Pass Pharmaceuticals, Inc. Vancouver, British Columbia, Canada.
   Prime Biotech, Paris, France
   EGene International, Forest City, CA
   Allowance: October 2002

8. "Defects in *MKKS* related to obesity, retinal dystrophy and renal malformations associated with Bardet-Biedl syndrome"
   Filed with United States Patent and Trademark Office: August 29, 2000
   Inventors: Nicholas Katsanis, Richard A. Lewis, James R. Lupski (Baylor College of Medicine)
   Phillip L. Beales (University College, London, UK)
   Michael O. Woods, Jane S. Green, Patrick S. Parfrey, William S. Davidson (University of Newfoundland, Canada)
   Licensee: Pending

9. "Periaxin mutations cause Charcot-Marie-Tooth disease and related neuropathies"
   Filed with United States Patent and Trademark office: December 13, 2000;
   Filed PCT Application in Canada and Japan: PCT/US01/48935, December 13, 2001
   Inventors: James R. Lupski, Cornelius F. Boerkoel III, Hiroshi Takashima
   Licensee: Athena Diagnostics, Worcester, MA.

10. "Molecular diagnosis of Smith-Magenis or del(17)(p11.2) microdeletion syndrome"
    Inventors: Pragna I. Patel, James R. Lupski, Frank Greenberg
    Licensee: Oncor, Gaithersburg, MD.

11. "*FLI* probes for FISH detection of SMS deletion"
    Inventors: James R. Lupski, Lisa G. Shaffer, Ken-Shiung Chen, Hugh D. Campbell
    Licensee: CytoCell Limited, Oxfordshire, United Kingdom

12. "*PLP* probes for FISH detection of Pelizaeus-Merzbacher disease (PMD) duplications"
    Inventors: James R. Lupski, Lisa G. Shaffer, Ken Inoue, A.S. Spikes
    Licensee: Pending

13. "*EGR2* transcription factor gene mutations associated with human myelinopathies"
    Inventors: James R. Lupski, Laura E. Warner, Cornelius Boerkoel
    Licesnsee: Athena Diagnostics, Worcester, MA.

14. "*SOX10* and dysmyelinating disease"
    Inventors: James R. Lupski and Ken Inoue
    Licensee: Pending

15. "An *in silico* subtraction method for the identification of tissue-specific genes"
Copyright: Nicholas Katsanis, Kim C. Worley, James R. Lupski and Pamela Culpepper
Filed with Register of Copyrights, Library of Congress: November 8, 1999
United States Copyright: TXu1-005-729; Registered May 11, 2001
Licensee: Pending

TEXAS CHILDREN'S HOSPITAL GENERAL CLINICAL RESEARCH CENTER PROTOCOLS:

1. A Correlated Clinical, Cytogenetic, and Molecular Genetic Analysis of del(17)(p11.2) Patients with Smith-Magenis Syndrome; 1990-2000.
P.I.: James R. Lupski, M.D./Ph.D.
Associate Investigators: Lorraine Potocki, M.D.; Richard A. Lewis, M.D., M.S.; Daniel Glaze, M.D.; Frank Brown, M.D., Ellen Friedman, M.D., Marcelle Sulek, M.D.
2. Construction of a Human Peripheral Nerve Specific cDNA Library to Identify Candidate Charcot-Marie-Tooth Disease Genes and Establish a Peripheral Nerve EST Database; 1990-present.
P.I.: James R. Lupski, M.D./Ph.D.
Associate Investigator: Dawna L. Armstrong, M.D.

GRANT REVIEWER:

National Science Foundation, International Programs
Veteran's Administration, Medical Research Service, Merit Review System
National Institutes of Health, ad hoc Review Committee for Bacteriology and Mycology-1 Study Section
National Institute of Neurological Disorders and Stroke, N.I.H. ad hoc for Neuroscience Program Project Grants
National Science Foundation, ad hoc for Microbial Genetics
Israeli Science Foundation
Medical Research Council, Canada, ad hoc reviewer for Human Genetics
Belgian National Fund for Scientific Research (NFSR)
Italian Telethon
Biotechnology and Biological Sciences Research Council, United Kingdom
The Wills Foundation, Houston, Texas
National Institutes of Health, Medical Scientist Training Program ad hoc for site visit
Muscular Dystrophy Association of Canada
University of Antwerp, Belgium, Special Research Fund-New Research Initiatives.
The Netherlands Organization for Scientific Research (CMR-NWO), Council for Medical Research
National Eye Institute, N.I.H. ad hoc for Clinical Research Study Section
The Wellcome Trust, United Kingdom, ad hoc for Training Fellowships
Medical Research Council, United Kingdom, ad hoc for Senior Non-Clinical Fellowship
National Institutes of Health, ad hoc Mammalian Genetics Study Section
Foundation Fighting Blindness
March of Dimes Birth Defects Foundation
National Institutes of Health, ad hoc Visual Sciences A Study Section
Association Francais contre les Myopathies (AFM)
Human Frontier Science Program
Medical Research Council, United Kingdom. ad hoc for Molecular and Cellular Medicine Board
United States – Israel Binational Science Foundation
The Fund for Scientific Research – Flanders, Belgium (FWO)
The Wellcome Trust, United Kingdom, ad hoc for Molecular and Cell Biology Grant

Austrian Science Fund, Hertha-Firnberg Fellowships
National Institutes of General Medical Sciences, N.I.H. Special Emphasis Panel
Innovation Oriented Research Program (IOP) on Genomics, The Netherlands.
Civilian Research and Development Foundation (CRDF), Science Center Programs of the U.S. Department of State
Italian Ministry for University and Research
The Netherlands Organization for Scientific Research (NWO) special program in genomics research.
National Institute of Neurological Disorders and Stroke ad hoc for Special Emphasis Panel ZNS1 SRB-5
Familial Dysautonomia Foundation
The Hospital for Sick Children Foundation. Toronto, Ontario, Canada.
Genome Canada

NIH STUDY SECTION MEMBER:

Biomedical Research Training – II, 2000-2003.

PEER REVIEWER:

American Journal of Clinical Investigation
American Journal of Human Genetics
American Journal of Medical Genetics
Annals of Neurology
Biochimica et Biophysica Acta
BioMed Central
BioTechniques
Brain
Canadian Journal of Microbiology
Circulation
Clinical Chemistry
Clinical Genetics
Clinical Infectious Diseases
European Journal of Human Genetics
European Journal of Infectious Diseases
Gene
Genetics in Medicine
Genome Biology
Genome Research
Genomics
Human Genetics
Human Molecular Genetics
Human Mutation
Infection Control and Hospital Epidemiology
Investigative Ophthalmology and Visual Sciences (IOVS)
Journal of the American Medical Association
Journal of Bacteriology
Journal of Biological Chemistry
Journal of Clinical Investigation
Journal of Infectious Diseases
Journal of Medical Genetics

Journal of Neuroscience
Journal of Neuroscience Research
Journal of the Peripheral Nervous System
Lancet
Mammalian Genome
Mayo Clinic Proceedings
Mental Retardation and Developmental Disabilities Research Reviews
Microbiology and Molecular Biology Reviews
Microscopy Research and Technique
Molecular Biology and Evolution
Molecular and Cellular Neuroscience (MCN)
Molecular and Cellular Probes
Mutation Research
Nature Reviews Genetics
Nature Genetics
Neurobiology of Disease
Neurogenetics
Neuron
Neuroscience Letters
Oncogene
Pediatrics
PLoS Biology
Prenatal Diagnosis
Proceedings of the National Academy of Sciences U.S.A.
PCR Methods and Applications
Science
Trends in Genetics

PATENT EVALUATOR:

Expert Opinion in Therapeutic Patents

PROFESSIONAL MEMBERSHIPS:

Genetics Society of America
American Association for the Advancement of Science
American Society of Human Genetics
American Society for Microbiology
American Academy of Pediatrics
American Federation for Medical Research
Harris County Hospital Society
Texas Medical Association
Southern Medical Association
New York Academy of Sciences
American Medical Association
New York State Medical Society
United States Chess Federation
New York State Chess Association

PROFESSIONAL SOCIETY ACTIVITIES:

Co-organizer and Co-convener with Dr. George Weinstock of session entitled, "Analysis of prokaryotic genomes" at the American Society for Microbiology, 1992 Annual Meeting, New Orleans, Louisiana, May 1992.

Co-organizer and Co-convener with Dr. Akio Ohnishi of Workshop No. 26 entitled, "Molecular genetics of inherited peripheral neuropathies including Charcot-Marie-Tooth disorders" at The VIII International Congress on Neuromuscular Diseases, Kyoto, Japan, July 1994.

Abstract reviewer, American Society of Human Genetics 1995 Annual Meeting.

Co-chair with Professor Dr. Christine van Broeckhoven of the 5th European CMT Consortium, London, England, April 14, 1996.

Nominating Committee for Board of Directors of the American Board of Medical Genetics, 1997.

Organizer of Symposium entitled "Region specific repeats and chromosomal rearrangements." American Society of Human Genetics, 1998 Annual Meeting, Denver, Colorado, October 1998.

Abstract reviewer, American Society of Human Genetics 2001 Annual Meeting.

Oral History Committee, American Society of Human Genetics, 2002-present

ELECTED SOCIETIES:

Society for Pediatric Research (Elected 1992)
American Society for Clinical Investigation (Elected 1998)

SERVICE:

Departmental

Glassware Facility Committee 1990-present
Promotions Committee 1995-present
Chairman Advisory Committee 1995-present
Diagnostic Laboratory Committee 1995-2000
Chair, Physician-Scientist Search Committee,
Department of Molecular and Human Genetics 1995-1998
Departmental Seminar Committee 1999-2001
Chair, Physician-Scientist Search Committee
Department of Molecular and Human Genetics 2001

College

Faculty Search Committee, Ophthalmology Department 1994 - 1995
Advisory Committee on Research Ethics, Center for Medical
Ethics and Health Policy 1995-1997
Search Committee, Director of Center for Gene Therapy 1995-1996
Program Development Committee for Baylor College of Medicine Vision 2000, 1996.
Internal Scientific Review Committee for Department of Obstetrics and Gynecology, 1997.

Pediatric Research Committee, Department of Pediatrics, 1997-1998
Departmental Research Advisory Committee, Department of Otorhinolaryngology and Communicative Sciences, 1997-1999.
Faculty Appointments and Promotions Committee, 1997-present.
Advisory Board Children's Health Research Center, 1997-present.
Selection Committee for Michael E. DeBakey, M.D. Excellence in Research Awards; 1999, 2001.
Incentives and Compensation Principles Task Planning Group, 1999
Statistical Genetics Recruitment Committee, 2000
Distinguished Educator Award Committee, 2000-present
Department Planning and Chair Review Committee 2001-present.

CLINICAL RESPONSIBILITIES/PRIVELEGES (1991-present; clinical sabbatical 1998-1999 academic year)

Prenatal Genetics consultation service, 8-10 weeks/year (1991-1997)
General Pediatrics ward attending at Ben Taub General Hospital, 1 month/year (1991-1998)
Neurofibromatosis Multidisciplinary Clinic, ½ day/month (1991-2000)
Genetics In-patient consultation service, 5-8 weeks/year (1991-present)
General Genetics Clinic, Texas Children's Hospital ½ day/week (1991-present)
General Genetics Clinic, Ben Taub General Hospital ½ day/month (1991-present)

HONORS:

Institute of Medicine of the National Academies of Science. Elected Member October 2002.

Curt Stern Award 2002, American Society for Human Genetics recognizing most significant achievement in human genetics in previous decade for delineating concept of genomic disorders.

John B. Carter, Jr. Family Tehnology Innovation Award 2002, for cofounding Bacterial BarCodes, Inc. based on the patented rep-PCR bacterial DNA fingerprinting technology.

Outstanding Investigator Award in Clinical Science for 2001, American Federation for Medical Research, for studies on the molecular genetics of inherited neuropathies.

The Solomon A. Berson Medical Alumni Achievement Award in Basic Science, New York University School of Medicine, April 2000.
This award is named since 1979 for Dr. Solomon A. Berson, NYU class of '45, whose contributions to the development of radioimmunoassay have led to enormous progress in basic sciences and clinical disciplines. Given since 1954 previous awardees include Jonas Salk, Albert Sabin and Eric Kandel.

Sackler Visiting Professor, Tel Aviv University, Israel, November 1999.

E. Mead Johnson Award for Pediatric Research 1998, The Society for Pediatric Research, for investigation of the molecular genetics of Charcot-Marie-Tooth disease and related demyelinating neuropathies.
Awarded since 1939 to honor laboratory and clinical research achievements in pediatrics. Previous awardees include Albert Sabin and Benjamin Spock.

DeBakey Research Award 1997 coawardee Dr. Richard A. Lewis for investigation of the molecular genetics of macular degeneration.

Fellow, American Association for the Advancement of Science, elected September 20, 1996.

Alpha Omega Alpha (AOA) New York University School of Medicine Outstanding Alumni Award, 1995.

Endowed Chair, Cullen Professor, in the Department of Molecular and Human Genetics, 1995-present.

Charcot-Marie-Tooth Association: The Distinguished Research Award for Outstanding Contributions to the Understanding of the Genetics of Charcot-Marie-Tooth Disorders. October 2, 1993.

American Federation for Clinical Research (AFCR) Southern Section Young Clinical Investigator Award, 1991

PEW Scholar in Human Genetics 1990-1994

Abbott Laboratories, Young Investigator of the Year Award 1989

Young Investigator Award, American Society for Microbiology Interscience Conference on Antimicrobial Agents and Chemotherapy (ICAAC) 1988

Inducted into the Hicksville Hall of Fame, June 21, 1987

TEACHING EXPERIENCE:

Undergraduate course lecturer in: Inorganic Chemistry, Cell Biology.

Graduate course lecturer in: Molecular Biology, Scientific Journalism, Human Genetics, Prokaryotic Genetics, Medical Genetics, Advanced Topics in Genetics.

Course coordinator for "Prokaryotic Genetics" course in Baylor College of Medicine Graduate School 1991-1993 and "Microbial Genetics" 1994-1996.

Baylor Laboratory Training Program Workshop, Lecturer 1993, 1994.

Baylor Pediatric Postgraduate Symposium 1994.

High School for the Health Professions Annual Science Symposium "Implications of the human genome project for patient care and the practice of medical genetics" 1996.

Introduction to Academic Medicine, Lecture course for Clinical Fellows in Department of Pediatrics. Course lecturer on Molecular Biology 1996-2000.

POSTDOCTORAL TRAINEES:

Past Trainees	Years	Independent Funding	Current P sition
Brunella Franco, M.D. (co-sponsored with Dr. P.I. Patel)	1989-1991	Muscular Dystrophy Assoc. (MDA)	Sr. Scientist, Telethon Institute of Genetics & Medicine (TIGEM), Milano, Italy

Vito Guzzetta, M.D. (co-sponsored with Dr. P.I. Patel)	1990-1991	Muscular Dystrophy Assoc. (MDA)	Sr. Scientist, Dept. of Pediatrics, Federico II University, Naples, Italy
Carol Wise, Ph.D.	1991-1992		Asst. Prof., UT-SW, Dallas, TX
Charles Woods, M.D.	1991-1992		Assoc. Prof., Wake Forest University School of Medicine, Winston-Salem, NC
Paul Georghiou, M.D.	1991-1993		Asst. Prof., Wesley Medical Center, Auchenflower, Queensland, Australia
Nacer Abbas, Ph.D.	1992-1994	Muscular Dystrophy Assoc. (MDA)	Groupe Hopitalier Pitié-Salpêtrière, Paris, France
Benjamin Roa, Ph.D.	1991-1994	Muscular Dystrophy Assoc. (MDA)	Asst. Prof., Molecular & Human Genetics, BCM, Houston, TX
Tatsufumi Murakami, M.D., Ph.D.	1994-1996	Muscular Dystrophy Assoc. (MDA)	Assoc. Prof., Kumamoto University School of Medicine
Qi Zhao, Ph.D.	1996-1997		Postdoctoral Fellow, NIH, NCI, Bethesda, MD
Yixin Li, M.D., Ph.D.	1996-1998		Scientist, Applied Biosystems
Ken-Shiung Chen, Ph.D.	1992-1998		Asst. Prof., University of Singapore, Taiwan, China
Lawrence Reiter, Ph.D.	1997-1999	Charcot-Marie-Tooth Assoc. (CMTA)	Postdoctoral Fellow, UCSD, San Diego, CA
Lorraine Potocki, M.D.	1995-1999	NIH NICHD (K08 HD01149)	Asst. Prof., Molecular & Human Genetics, BCM, Houston, TX
Bassem Bejjani, M.D.	1995-1999	NIH NRSA NIH NEI (K08 EY00375)	Res. Prof., Washington State University, Spokane, WA
Sung Sup Park, M.D., Ph.D.	1999-2000	Korean Fellowship	Prof., Seoul National University
David Stockton, M.D.	1995-2001	Daland Fellowship & Am. Philosophical Society Knight Templar Eye Fnd NIH NEI (K08 EY00375)	Asst. Prof., Molecular & Human Genetics, BCM, Houston, TX
Cornelius F. Boerkoel, III, M.D., Ph.D.	1998-2001	NIH NIDDK (K08 DK02738)	Asst. Prof., Molecular & Human Genetics, BCM, Houston, TX
Nicholas Katsanis, Ph.D.	1997-2002	March of Dimes	Asst. Prof., Johns Hopkins School of Medicine, Baltimore, MD
Hiroshi Takashima, M.D., Ph.D.	2000-2002	Charcot-Marie-Tooth Assoc. (CMTA)	Asst. Prof., Kagoshima University, Kagoshima, Japan
Keiko Wakui, Ph.D.	**2002-2003**		Division of Medical Genetics, Shinshu University School of Medicine, Asahi, Matsumoto, Nagano, Japan

Current Trainees	Years	**Independent Funding**
Ken Inoue, M.D., Ph.D.	1997-1999 2000-present	Charcot-Marie-Tooth Assoc. (CMTA) Muscular Dystrophy Association postdoctoral fellowship

Katherina Walz, Ph.D.	1999-present	
Alexander Yatsenko, Ph.D.	1999-present	Fight for Sight, Prevent Blindness America
Pawel Stankiewicz, M.D., Ph.D.	1999-present	
Weimin Bi, Ph.D.	2000-present	
G. Mustafa Saifi, Ph.D.	2001-present	Charcot-Marie-Tooth Association (CMTA)
Kinga Szigeti, M.D.	2002-present	
Wojciech Wiszniewski, M.D., Ph.D.	2003-present	
Naohiro Kurotaki, M.D., Ph.D.	2003-present	

PREDOCTORAL TRAINEES:

James Versalovic; Cell and Molecular Biology Program and N.I.H. Medical Scientist Training Program 1989-1994; Ph.D. 1994, M.D. 1995. "Evolution of the macromolecular synthesis operon and analysis of bacterial primase".

N.I.H. individual predoctoral fellowship.
Winner of The BF Goodrich Collegiate Inventors Program, 1992 Inventor of the Year. Student/Advisor Award ($7,500) for invention entitled, "Fingerprinting microorganisms using repetitive DNA sequence amplification".
Winner of the 1993 Sheard Sanford Medical Student Award from the American Society of Clinical Pathologist.
Current Position: Assistant Professor, Baylor College of Medicine

Kent Anderson; Institute for Molecular Genetics and N.I.H. Medical Scientist Training Program 1990-1996; Ph.D. 1996, M.D. 1998. "Towards the isolation of genes for recessively inherited ocular disorders: Bardet-Biedl syndrome, Leber congenital amaurosis, primary congenital glaucoma, and Stargardt disease".

N.E.I. predoctoral training grant fellowship.
Current Position: unknown

Robert Britton; Cell and Molecular Biology Program, 1991-1996; Ph.D. 1996. "Suppressor analysis of *E. coli dnaG* mutations".

Winner of 1994 O.B. Williams Award, Texas Branch American Society for Microbiology
Current Position: Assistant Professor, Michigan State University

Larry Reiter; Cell and Molecular Biology Program, 1994-1998; Ph.D. 1998. "Homologous recombination at the CMT1A duplication/HNPP deletion locus: Mapping of a hotspot for unequal crossing over events."
1996 Boehringer Ingelheim Fonds Travel Award to perform experiments for two months in Dr. Bernd Rautenstrauβ's laboratory at the Institute of Human Genetics, Erlangen-Nürnberg, Germany and two weeks at Prof. Dr. Christine van Broeckhoven's Neurogenetics Laboratory of the University of Antwerpen, Belgium.
Claude W. Smith award for outstanding research performance in the Cell and Molecular Biology Program, 1996.
Sigma Xi Ph.D. thesis award, 1998.
Current Position: Postdoctoral Fellow, UCSD (Ethan Bier)

Laura Warner; Department of Molecular and Human Genetics, 1994-1999, Ph.D. 1999. "Mutational analysis and elucidation of the molecular mechanisms resulting in human myelinopathies."
N.I.G.M.S. predoctoral training grant.
Current Position: Research Scientist, University of Washington

Noah Shroyer; Cell and Molecular Biology Program, 1996-2001, Ph.D. 2001. "The photoreceptor-specific ATP-binding cassette gene, ABCR, in Mendelian and multifactorial retinal disease."
N.E.I. predoctoral training grant fellowship.
Claude W. Smith Award for outstanding research performance in the Cell and Molecular Biology Programs, 1999.
Current Position: Postdoctoral Fellow, Baylor College of Medicine (Huda Zoghbi)

Jiong Yan; Department of Molecular and Human Genetics, 1999-.

Erica Eichers; Department of Molecular and Human Genetics, 2000-.

Jose Badano; Department of Molecular and Human Genetics, 2000-.
Fullbright Scholarship

Christine J. Shaw; Department of Molecular and Human Genetics, 2001-.

Mehrdad Khajavi; Department of Molecular and Human Genetics, 2001-.

Jennifer Lee; Department of Molecular and Human Genetics, 2002-.

Patricia Fonseca; Department of Molecular and Human Genetics, 2002-.

Gabriel Bien-Willner; Department of Molecular and Human Genetics, Medical Scientist Training Program, 2003-.

MEDICAL STUDENT FELLOW:

Charles M. Zaremba; Howard Hughes Research Training Fellowship for Medical Students, 2003-2004.

THESIS COMMITTEES:

Participated in Ph.D. committees of 16 graduates and of 15 current students.

BIBLIOGRAPHY:

(A.) Original Articles:

1981

1. Chaconas, G., de Bruijn, F.J., Casadaban, M., and **Lupski, J.R.**, Kwoh, T.J., Harshey, R.M., DuBow, M.S., and Bukhari, A.I. (1981). *In-vitro* and *in-vivo* manipulations of bacteriophage Mu DNA: Cloning of Mu ends and construction of mini Mu's carrying selectable markers. *Gene 13*: 37-46.

1982

2. **Lupski, J.R.**, Smiley, B.L., Blattner, F.R., and Godson, G.N. (1982). Cloning and characterization of the *Escherichia coli* chromosomal region surrounding the *dnaG* gene, with a correlated physical and genetic map of *dnaG* generated via transposon Tn*5* mutagenesis. *Molec. Gen. Genet. 185*:120-128.
3. Smiley, B.L., and **Lupski, J.R.**, Svec, P., McMacken, R., Godson, G.N. (1982). Sequences of the *Escherichia coli dnaG* primase gene and regulation of its expression. *Proc. Natl. Acad. Sci. USA 79*:4550-4554.

1983

4. **Lupski, J.R.**, Smiley, B.L., and Godson, G.N. (1983). Regulation of the *rpsU-dnaG-rpoD* macromolecular synthesis operon and the initiation of DNA replication in *Escherichia coli* K-12. *Molec. Gen. Genet. 189*:48-57.
5. **Lupski, J.R.**, Ozaki, L.S., Ellis, J., and Godson, G.N. (1983). Localization of a *Plasmodium* surface antigen epitope by Tn*5* mutagenesis mapping of a recombinant cDNA clone. *Science 220*:1285-1288.

1984

6. **Lupski, J.R.**, Gershon, P., Ozaki, L.S., and Godson, G.N. (1984). Specificity of Tn*5* insertions into a 36 nucleotide DNA sequence repeated in tandem seven times. *Gene 30*:99-106.

7. **Lupski, J.R.**, Ruiz, A.A., and Godson, G.N. (1984). Promotion, termination, and anti-termination in the *rpsU-dnaG-rpoD* macromolecular synthesis operon of *E. coli* K-12. *Molec. Gen. Genet. 195*:391-401.

8.

1985

9. Peacock, S., **Lupski, J.R.**, Godson, G.N. and Weissbach, H. (1985). *In vitro* stimulation of *Escherichia coli* RNA polymerase sigma subunit synthesis by NusA protein. *Gene 33*:227-234.
10. Bukhari, A.I., and **Lupski, J.R.**, Svec, P. and Godson, G.N. (1985). Comparison of left-end DNA sequences of bacteriophages Mu and D108. *Gene 33*:235-239.

1986

11. Cudny, H., **Lupski, J.R.**, Godson, G.N. and Deutscher, M.P. (1986). Cloning, sequencing and species relatedness of the *Escherichia coli cca* gene encoding the enzyme tRNA nucleotidyltransferase. *J. Biol. Chem. 261*:6444-6449.

12. **Lupski, J.R.**, Projan, S., Ozaki, L.S. and Godson, G.N. (1986). A temperature dependent pBR322 copy number mutant resulting from a Tn*5* position effect. *Proc. Natl. Acad. Sci USA, 83*:7381-7385.

1987

13. Nesin, M., **Lupski, J.R.**, Svec, P. and Godson, G.N. (1987). Possible new genes as revealed by molecular analysis of a 5.0 kb *E. coli* chromosomal region 5' to the macromolecular synthesis operon. *Gene 55*:149-161.

1988

14. Nesin, M., **Lupski, J.R.**, and Godson, G.N. (1988). Role of 5' upstream sequences and the tandem promoters in regulation of the *rpsU-dnaG-rpoD* macromolecular synthesis operon. *J. Bacteriol. 170*:5759-5764.

1990

15. Patel, P.I., Franco, B., Garcia, C., Slaugenhaupt, S.A., Nakamura, Y., Ledbetter, D.H., Chakravarti, A., and **Lupski, J.R.** (1990). Genetic mapping of autosomal dominant Charcot-Marie-Tooth disease in a large French-Acadian Kindred: Identification of new linked markers on chromosome 17. *Am. J.Hum. Genet. 46*:801-809.
16. **Lupski, J.R.**, Zhang, Y.H., Rieger, M., Minter, M., Shu, B., Doi, B.G., Koeuth, T. and McCabe, E.R.B. (1990). Mutational analysis of the *E. coli glpFK* region with Tn*5* mutagenesis and the polymerase chain reaction. *J. Bacteriol. 172*:6129-6134.
17. Nesin, M., Svec, P., **Lupski, J.R.**, Godson, G.N., Kreiswirth, B. and Projan, S.J. (1990). Cloning and nucleotide sequence of a chromosomally encoded tetracycline resistance determinant, *tetA*(M), from a pathogenic, methicillin-resistant strain of *Staphylococcus aureus*. *Antimicrobial Agents and Chemotherapy 34*: 2273-2276.
18. Patel, P.I., Garcia, C., Montes de Oca Luna, R., Malamut, R., Franco, B., Slaugenhaupt, S., Chakravarti, A. and **Lupski, J.R.** (1990). Isolation of a marker linked to the Charcot-Marie-Tooth disease type 1A a gene by differential *Alu* PCR of human chromosome 17-retaining hybrids. *Am. J. Hum. Genet. 47*:926-934.

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19. Guzzetta, V., Montes de Oca-Luna, R., **Lupski, J.R.**, and Patel, P.I. (1991). Isolation of region-specific and polymorphic markers from chromosome 17 by restricted *Alu*-PCR. *Genomics 9*:31-36.
20. Grompe, M., Versalovic, J., Koeuth, T., and **Lupski, J.R.** (1991). Mutations in the *Escherichia coli dnaG* gene suggest coupling between DNA replication and chromosome partitioning. *J. Bacteriol. 173*:1268-1278.
21. **Lupski, J.R.**, Montes de Oca-Luna, R., Slaugenhaupt, S., Pentao, L., Guzzetta, V., Trask, B.J., Saucedo-Cardenas, O., Barker, D.F., Killian, J.M., Garcia, C.A., Chakravarti, A., and Patel, P.I. (1991). DNA duplication associated with Charcot-Marie-Tooth disease type 1A. *Cell* 66:219-232.
22. Franco, B., Lai, Li-Wen, Patterson, D, Ledbetter, D.H., Trask, B.J., van den Engh, G., Iannaccone, S., Frances, S., Patel, P.I. and **Lupski, J.R.** (1991). Molecular characterization of a patient with del(1) q23-q25. *Hum. Genet. 87*:269-277.
23. **Lupski, J.R.**, Garcia, C.A., Zoghbi, H.Y., Hoffmann, E.P., and Fenwick, R.G. (1991). Discordance of muscular dystrophy in monozygotic female twins: Evidence supporting asymmetric splitting of the inner cell mass in a manifesting carrier. *Am. J. Med. Genet. 40*:354-364.
24. Versalovic, J., Keouth, T., McCabe, E.R.B. and **Lupski, J.R.** (1991). Use of the polymerase chain reaction (PCR) for physical mapping of *E. coli* genes. *J. Bacteriol. 173*:5253-5255.

25. Hoffman, E.P., Garcia, C.A., Chamberlain, J.S., Angelini, C., **Lupski, J.R.**, and Fenwick, R.G. (1991). Is the carboxyl-terminus of dystrophin required for membrane association? A novel, severe case of Duchenne muscular dystrophy. *Annals of Neurology 30*:605-610.
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1992

28. Subramanian, P., Versalovic, J., McCabe, E.R.B. and **Lupski, J.R.** (1992). Rapid mapping of *E. coli*::Tn*5* insertion mutations by REP-Tn*5* PCR. *PCR Methods and Applications 1*:187-194.
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32. Guzzetta, V., Franco, B., Zhang, H., Saucedo-Cardenas, O., Montes de Oca-Luna, R., Greenberg, F., Chinault, A.C., **Lupski, J.R.** and Patel, P.I. (1992). Somatic cell hybrids, sequence tagged sites, simple repeat polymorphisms and yeast artificial chromosomes for physical and genetic mapping of proximal 17p. *Genomics 13*:551-559.
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1993

36. **Lupski, J.R.**, Pentao, L., Williams, L.L. and Patel, P.I. (1993). Stable inheritance of the CMT1A DNA duplication in two patients with CMT1 and NF1. *Am. J. Med. Genet 45*:92-96.
37. Versalovic, J., Kapur, V., Mason, E.O., Jr., Shah, U., Koeuth, T., **Lupski, J.R.**, and Musser, J.M. (1993). Penicillin-resistant *Streptococcus pneumoniae* strains recovered in Houston: Identification and molecular characterization of multiple clones. *J. Infect. Dis., 167*:850-856.
38. Versalovic, J., Koeuth, T., Britton, R., Geszvain, K., and **Lupski, J.R.** (1993). Conservation and evolution of the *rpsU-dnaG-rpoD* macromolecular synthesis operon in eubacteria. *Molecular Microbiology 8*:343-355.
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42. Zori, R.T., **Lupski, J.R.**, Zhang, H., Greenberg, F., Killian, J.M., Gray, B.A., Driscoll, D.J., Patel, P.I., and Zackowski, JL. (1993). An infant with Smith-Magenis syndrome born from a mother having a mosaic 17p11.2p12 deletion. *Am. J. Med. Genet. 47*:504-511.
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47. Versalovic, J. and **Lupski, J.R.** (1993). The *Haemophilus influenzae dnaG* sequence and conserved bacterial primase motifs. *Gene 136*:281-286.

1994

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49. Leppert, M., Baird, L., Anderson, K., Otterud, B., **Lupski, J.R.**, and Lewis, R.A. (1994). Bardet-Biedl syndrome links to DNA markers on chromosome 11q and is genetically heterogeneous. *Nature Genetics 7*:108-112.
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51. Shapira, S.K., Anderson, K., Orr-Urtreger, A., Craigen, W.J., **Lupski, J.R.**, and Shaffer, L.G. (1994). *De novo* proximal deletions of 14q: Clinical, cytogenetic and molecular investigations. *Am. J. Med. Genet. 52*:44-50.
52. Suter, U., Snipes, G.J., Schoener-Scott, R., Welcher, A.A., Pareek, S., **Lupski, J.R.**, Murphy, R.A., Shooter, E.M., and Patel, P.I. (1994). Regulation of tissue-specific expresssion of alternative peripheral myelin protein-22 (*PMP22*) gene transcripts by two promoters. *J. Biol. Chem. 269*:25795-25808.
53. Georghiou, P.R., Doggett, A.M., Kielhofner, M.A., Watson, D.A., **Lupski, J.R.**, and Hamill, R.J. (1994). Molecular fingerprinting of *Legionella* species using repetitive element polymerase chain reactions (rep-PCR). *J. Clin. Microbiol. 32*:2989-2994.
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(C.) Invited Reviews

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340. Wakui, K., Inoue, K., Shaffer, L.G., and **Lupski, J.R.** (2003). Genomic inversion polymorphism in parents of patients with Williams-Beuren syndrome. 48th Annual Meeting of the Japan Society of Human Genetics, Nagasaki, JAPAN.
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360. Khajavi, M., Oyama, T., Inoue, K., and **Lupski, J.R.** (2003). Nonsense mediated decay mitigates the effect of *MPZ* mutant alleles responsible for a more severe peripheral neuropathies. Annual Meeting of the American Society of Human Genetics, Los Angeles, California. *Am. J. Hum. Genet.* 73:A2306.
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362. Fonseca, P., Walz, K., and **Lupski, J.R.** (2003). Identifying dosage-sensitive genes in the Smith-Magenis syndrome region. Annual Meeting of the American Society of Human Genetics, Los Angeles, California. *Am. J. Hum. Genet.* 73:A2350.
363. Saifi, G.M., Szigeti, K., Wiszniewski, W., and **Lupski, J.R.** (2003). Mutations in the *SIMPLE* gene are a relative common cause of dominant Charcot-Marie-Tooth disease (CMT1C) and related peripheral neuropathies. Annual Meeting of the American Society of Human Genetics, Los Angeles, California. *Am. J. Hum. Genet.* 73:A2419.
364. Stockton, D.W., Quijano, R., Nguyen, D., Leppert, M., Lewis, R.A., and **Lupski, J.R.** (2003). An exploration of Leber congenital amaurosis locus, allelic, and inheritance complexity. Annual Meeting of the American Society of Human Genetics, Los Angeles, California. *Am. J. Hum. Genet.* 73:A2466.
365. Potocki, L., Walz, K., Treadwell-Deering, D.E., Krull, K., Glaze, D., Paylor, R., and **Lupski, J.R.** (2003). Behavior and sleep disturbances in the 17p11.2 microduplication and microdeletion syndromes: Comparisons and contrasts between mouse and human. American College of Medial Genetics Annual Clinical Genetics Meeting, Kissimmee, Florida.
366. Wiszniewski, W., Zaremba, C.M., Yatsenko, A., Jamrich, M., Wensel, T., and **Lupski, J.R.** (2003). Expression of human *ABCR* in *Xenopus laevis* photoreceptors. 2004 Association for Research in Vision and Ophthalmology (ARVO), Fort Lauderdale, Florida.
367. Zaremba, C.M., Yatsenko, A., Wiszniewski, W., Leppert, M., Lewis, R., **Lupski, J.R.** (2003). *ABCA4* mutational analysis in a cohort of 30 recessive retinitis pigmentosa families. 2004 Association for Research in Vision and Ophthalmology (ARVO), Fort Lauderdale, Florida.
368. Yatsenko, A.N., Wiszniewski, W., Lewis, R.A., and **Lupski, J.R.** (2003). An evolutionary comparative analysis of abcr proteins in vertebrates' species. 2004 Association for Research in Vision and Ophthalmology (ARVO), Fort Lauderdale, Florida.

369. Lewis, R., Krajewski, K., Shy, R., Saifi, M., **Lupski, J.R.**, and Shy, M. (2003). Classic, but mild CMT1 phenotype in CMT1C family with Gly112Ser mutation in *LITAF/SIMPLE*. 56th Annual Meeting of the American Academy of Neurology, San Francisco, California.

INVITED LECTURES: (2002-2003; total of 212 from 1982-2003)

168. Neurology Grand Rounds, Albert Einstein College of Medicine, New York, New York. "Diagnosis, clinical management and molecular mechanisms for hereditary peripheral neuropathies." January 15, 2002.
169. Whitehead Institute for Genomic Research, Cambridge, Massachusetts. "Genome architecture, rearrangements, evolution and genomic disorders." January 22, 2002.
170. BioGen, Cambridge, Massachusetts. "*ABCR* (*ABCA4*) in monogenic and multifactorial disease: evidence from family studies supporting the activity vs. retinopathy phenotype hypothesis. January 23, 2002.
171. Cornell University, Weill Medical College, Pediatric Grand Rounds. New York, New York. "Genome architecture, rearrangements, evolution, and genomic disorders." January 24, 2002.
172. St. Louis University Medical School, Department of Neurology, St. Louis, Missouri. "Diagnosis, clinical management and molecular mechanisms for hereditary peripheral neuropathy." March 14, 2002.
173. Department of Human Genetics, University of Michigan, Ann Arbor, Michigan. "Genome architecture, rearrangements, evolution and genomic disorders." April 30, 2002.
174. Department of Molecular and Medical Genetics, Oregon Health Sciences University, Portland, Oregon. "Genome architecture, rearrangements, evolution and genomic disorders." May 8, 2002.
175. Grand Rounds, Department of Molecular and Medical Genetics, Oregon Health Sciences University, Portland, Oregon. "Diagnosis, clinical management, and molecular mechanisms for hereditary peripheral neuropathies." May 9, 2002.
176. The Institute for Genetic Medicine and the Neurogenetic Institute Distinguished Speaker Seminar Series, University of Southern California, Los Angeles, California. "Genome architecture, rearrangements, evolution, and genomic disorders." May 16, 2002.
177. 3rd International Conference on Smith-Magenis Syndrome, PRISMS (Parents and Researchers Interested in Smith-Magenis Syndrome), Denver, Colorado. "Our SMS Mice: What will they teach us?" July 6, 2002.
178. Xth International Congress on Neuromuscular Diseases, Vancouver CANADA. "Hereditary Sensory-Motor Neuropathies." July 10, 2002.
179. Pediatric Grand Rounds, Medical University of South Carolina, Charleston, South Carolina. "Diagnosis, clinical management and molecular mechanisms for hereditary peripheral neuropathy." July 20, 2002.
180. South Carolina Genetics Conclave Meeting, Charleston, South Carolina. "Genome architecture, rearrangements, evolution, and genomic disorders." July 20, 2002.
181. Genzyme, Framingham, Massachusetts. "*ABCR* (*ABCA4*) in monogenic and multifactorial disease: evidence from family studies supporting the activity vs. retinopathy phenotype hypothesis." October 3, 2002.
182. Education Session. Novel mutational mechanisms: Implication for identifying mutations and defining new disease gene candidates. 2002 Annual Meeting of the American Society of Human Genetics, Baltimore, Maryland. "Genome architecture, rearrangements and genomic disorders." October 16, 2002.
183. Molecular basis of Mendelian inheritance. 2002 Annual Meeting of the American Society of Human Genetics, Baltimore, Maryland. "Phenotypic consequence of gene dosage imbalance in chromosome engineered mouse models for del(17)p11.2 (Smith-Magenis syndrome) and its reciprocal duplication." October 18, 2002.
184. Constitutional chromosomal rearrangements and the evolution of the human genome. 2002 Annual Meeting of the American Society of Human Genetics, Baltimore, Maryland. "Genome architecture

and primate evolution of 17p11.2p12, rearrangements and genomic disorders." October 16, 2002.

185. 2002 Annual Meeting of the American Society of Human Genetics, Baltimore, Maryland. Curt Stern Award Address: "Genomic disorders: recombination based disease resulting from genome architecture." October 18, 2002.

186. Department of Human Genetics, University of Chicago. Chicago, Illinois. "Genomic disorders: recombination based disease resulting from genome architecture." October 24, 2002.

187. Pediatric Grand Rounds, University of Pittsburgh School of Medicine/Children's Hospital of Pittsburgh, Pittsburgh, Pennsylvania. "Diagnosis, clinical management and molecular mechanisms for hereditary Peripheral neuropathy." November 7, 2002.

188. Department of Human Genetics, Graduate School of Public Health, University of Pittsburgh School of Medicine, Pittsburgh, Pennsylvania. "Genome architecture, rearrangements, evolution and genomic disorders." November 8, 2002.

189. Department of Molecular Genetics, M.D. Anderson Cancer Center, Houston, Texas. "Genome architecture, rearrangements, evolution and genomic disorders." January 22, 2003.

190. Telethon Institue of Genetics and Medicine (TIGEM), Naples, ITALY. "*ABCR* (*ABCA4*) in monogenic and multifactorial disease: evidence from family studies supporting the activity vs. retinopathy phenotype hypothesis." February 4, 2003.

191. Scuola Europa di Medicina Moleculare, Facoltà di Medicina e Chirugia dell'Ateneo Frederico II, Naples, ITALY. "The MD/PhD Program in the USA." February 5, 2003.

192. Telethon Institue of Genetics and Medicine (TIGEM), Naples, ITALY. "Genome architecture, rearrangements, evolution and genomic disorders." February 5, 2003.

193. CEINGE Biotecnologie Avanzate S.c.a.r.l., Naples, ITALY. "Genome architecture, rearrangements, evolution and genomic disorders." February 6, 2003.

194. Neurology Grand Rounds, Tulane University School of Medicine, New Orleans, Louisiana. "Hereditary Sensory-Motor Neuropathies: molecular mechanisms and applications of genetic testing." February 10, 2003.

195. Center for Molecular Medicine and Genetics, and the Department of Neurology, Wayne State University, School of Medicine. Detroit, Michigan. "Genome architecture, rearrangements, evolution and genomic disorders." March 6, 2003.

196. North American CMT Consortium Inaugural Meeting. London, Ontario, CANADA. "Disturbance of muscle fiber type differentiation in congenital hypomyelinating neuropathy." March 8, 2003.

197. Invited Address, 30th Annual Meeting of the Texas Genetics Society. Austin, Texas. "Genomic disorders: recombination-based disease resulting from genome architecture." March 28, 2003.

198. Cellular and Molecular Mechanisms of Macular Degeneration. The Ruth and Milton Steinbach Fund, 10th Anniversary Symposium, New York, New York. "*ABCR* (*ABCA4*) activity versus retinal disease phenotype hypothesis." April 5, 2003.

199. Neurology Grand Rounds, University of Oklahoma College of Medicine. Oklahoma City, Oklahoma. "Hereditary Sensory-Motor Neuropathies: molecular mechanisms and applications of genetic testing." May 13, 2003.

200. LXVIII Cold Spring Harbor Symposium on Quantitative Biology. The Genome of *Homo Sapiens*. Cold Spring Harbor, New York. "Genomic disorders-Genome architecture results in susceptibility to DNA rearrangements causing common human traits." May 29, 2003.

201. 7th Annual Symposium on Advances in Laboratory Medicine. A symposium in Honour of Dr. Laurence E. Becker. Toronto, Ontario, CANADA. "Genomic architecture, rearrangement, evolution and genomic disorders." June 5, 2003.

202. Program in Molecular Medicine, The Hospital for Sick Children, University of Toronto Faculty of Medicine. Toronto, Ontario, CANADA. "Nonsense mediated decay and human disease." June 5, 2003.

203. Gordon Research Conference. Human Genetics and Genomics. Colby College, Waterville, Maine. "Genome architecture, rearrangements, evolution and genomic disorders." August 5, 2003.

204. 49th National Congress of Genetics, Brazilian Society of Genetics. Aguas de Lindoia, Sao Paulo, BRAZIL. "Genome architecture, rearrangements, and genomic disorders." September 17, 2003.
205. Neurology Grand Rounds, University of Rochester, Medical Center, Rochester, New York. "Diagnosis, clinical management, and molecular mechanisms for hereditary peripheral neuropathies." October 12, 2003.
206. Research Institute for Neurological Diseases and Geriatrics, Department of Neurology and Gerontology, Kyoto Prefectural University of Medicine, Kyoto, JAPAN. "Genome architecture, rearrangements, evolution and genomic disorders." October 17, 2003.
207. Third Department of Internal Medicine, Faculty of Medicine, Kagoshima University, Kagoshima, JAPAN. "The genomic basis of disease – Implications of the human genome project for neurological disorders." October 20, 2003.
208. 48th Annual Meeting of the Japan Society of Human Genetics, Nagasaki, JAPAN. "Genomic disorders – genome architecture results in susceptibility to DNA rearrangements causing common human traits." October 23, 2003.
209. The Alfred W. Bressler Vision Science Symposium, The Jewish Guild for the Blind. New York Academy of Sciences, New York, New York. "The Stargardt disease/macular degeneration story: A single gene clue to complex traits." October 31, 2003.
210. Clinical Genetics I, 2003 Annual Meeting of the American Society of Human Genetics, Los Angeles, California. "Variability in clinical phenotype despite common chromosomal deletion in Smith-Magenis syndrome." November 7, 2003.
211. Neurology Grand Rounds, Medical College of Georgia, Augusta, Georgia. "Hereditary Motor and Sensory Neuropathies: Molecular mechanisms and applications of genetic testing." November 13, 2003.
212. Department of Cellular and Structural Biology, University of Texas Health Science Center at San Antonio. "Genome architecture, rearrangements, evolution and genomic disorders." December 4, 2003.

A COMPLETE CV MAY BE FURNISHED UPON REQUEST